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Case Report

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Consequences of Proximal Femoral Focal Deficiency Case Report and Literature Review

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Abstract

Introduction: *Proximal femoral focal deficit it is believe a congenital condition with phenomena known as dysplastic hip with shortening or absent femur. Proximal femoral focal deficit is relatively uncommon. Estimate of this infrequent anomaly on radiographs can aid treat these cases better as early treatment could help in achieving satisfactory growth of the femur.*

Importance: *We report one of the rarest cases of patient underwent five different surgeries for correction of proximal femoral focal deficit starting from age of 11 years to 25 years of age and dealing with proximal femoral focal deficit complication.*

Case Presentation: *11 years with left lower limb shortening. The Patient was not following up with any clinic before. He was born via vaginal normal delivery as full-term baby with no postnatal complication. Upon examination, the patient had a shortening of the left lower limb of almost 6cm in comparison to the right side. x ray showed left hip proximal femur focal deficit. Our patient underwent 5 different surgical correction of proximal femoral focal deficit.*

Clinical Discussion:
Proximal femoral focal deficit is an extremely rare congenital anomaly with few case reports in the literature. Therefore, for us to have one case with a long follow-up that includes five surgeries done to the same patient, which is not described in the existing literature, is exceptional. No literature describe how to deal with complication of Proximal femoral focal deficit.

Conclusion: *The discovery of one cases of proximal femoral focal deficiency, a rare entity. Moreover, dealing with such rare case with it is compilation consider a very rare entity.*

Keywords: Limb lengthening, Proximal Femur Focal Deficiency, Shortening limb, Femur, Flexion contracture.

1. Introduction

Femoral insufficiency includes a huge variety of difference anomaly. starting from dysplastic hip with relative normal femur, shortening femur, and most severe form which is total absent of femur with only distal femoral condyles present [1]. Proximal femoral focal deficit (PFFD), likewise acknowledged as congenital proximal femoral deficit (CPFD) is an unusual congenital skeletal disorder well-defined by the incomplete nonappearance of the proximal femoral segment with shortening of the entire lower extremities [1,2]. The incidence varies from 1 case per 50,000 people to 1 case per 200,000 people [2]. At one time, Proximal Femoral Focal Deficiency turn out to be sorted with other disorders, like coxa vara and short bowed femurs, which led to misperception and misinterpretation. Later, as result of a more thorough understanding, numerous classification system have been created [3,4]. The origin cause of PFFD is unknown. There is a theory that links it to a genetic disorder. However, the primary ossification center defect was the main issue [1]. 60% of the PFFD cases is unilateral, and only 15% to 30% of the cases is bilateral. Moreover, most of the unilateral cases affect left lower limb [5]. PFFD has been seen to be linked to other musculoskeletal disorder like short tibia and fibula, patellar hypoplasia, limb malrotation and knee joint instability with absence of ACL or PCL [1]. Deformities of the contralateral upper and lower limbs may also exist [4]. Up to 70% of individuals with PFFD may also have additional anomalies,

such as cleft palate and clubfoot [6]. Specific management for PFFD variety from observation to surgical intervention, and in some cases, efficient amputation. In our study, we report a case of isolated congenital unilateral PFFD with fracture of shaft of femur, knee dislocation, patella hypoplasia, knee flexion contracture. The diagnosis of PFFD is made in early childhood and based mainly on conventional radiography. Our patient underwent 5 different surgeries for lengthening and correction of deformity in PFFD and it is complication. The main goal for this case to present the consequence of surgical treatment of PFFD with is complication and following up a patient with more than 10 years.

2. Case report

We present this case of a patient underwent five different surgeries for correction of PFFD starting from of 11 years to 25 years of age. Our patient was referred to the pediatric orthopedic clinic at the age of 11 years with left lower limb shortening. The Patient was not following up with any clinic before. He was born via vaginal normal delivery as full-term baby with no postnatal complication.

Upon examination, the patient had a shortening of the left lower limb of almost 6cm in comparison to the right side. No swelling within the hips and no skin changes in the area. He had a decreased left hip Range of motion in flexion internal and external rotations. The left lower limb was short and he used a high shoe to compensate for shortening (Figure 1). Full-length radiographs of the lower limb and pelvic show features of PFFD (Figure 2).

Figure 1 : Shortening of left lower limb with compensation of high shoes

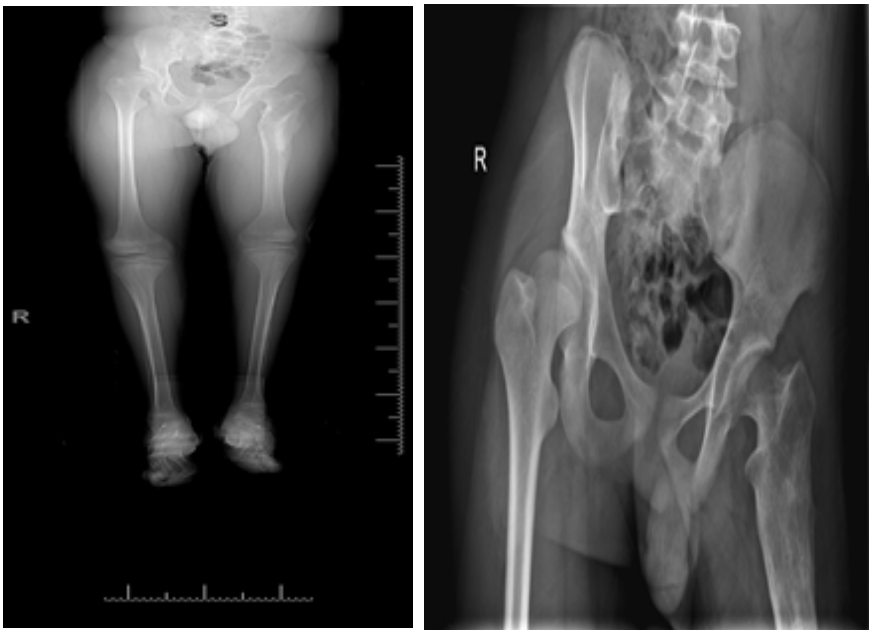


Figure 2: =X ray full length lower limb show: dysplastic of right and left acetabulum with proximal femur head deficiency, left proximal femur varus, distal femur condylar deficiency, shortening left lower limb

The patient at that time underwent left femur osteotomy and application of femur Taylor Spatial Frame (TSF) for lengthening (Figure 3) . He was followed for a long time, and we achieved almost 6cm lengthening. Serial radiographs were taken during lengthening, which showed complete healing of the osteotomy site and lengthening of the femur (Figure 4). One complication was encountered during lengthening, which is flexion contracture of the left knee, then we stopped lengthening after the removal of TSF patient was doing good functional daily activity.

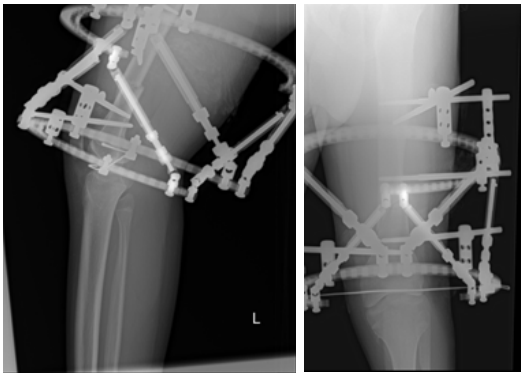
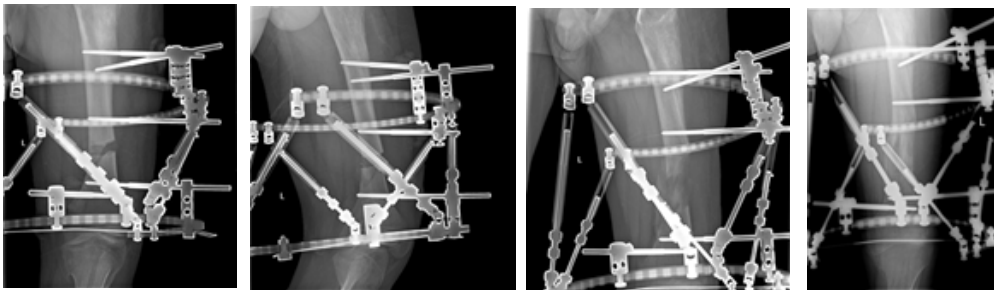


Figure 3: AP and lateral view X ray left femur and knee show distal femur osteotomy with application of TSF



AP and lateral view X ray left femur and knee show distraction and lengthening was initiated and achieved.

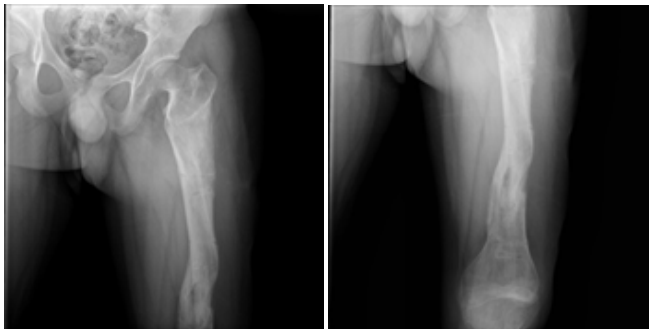


Figure 4: AP x ray of left femur show healing of lengthening osteotomy without hip or knee dislocation

At the age of 14 years, the patient had a simple fall and got a left knee dislocation (Figure 5). For this, he underwent a close reduction of knee dislocation followed by the application of spanning external fixation "TSF" (Figure 6).

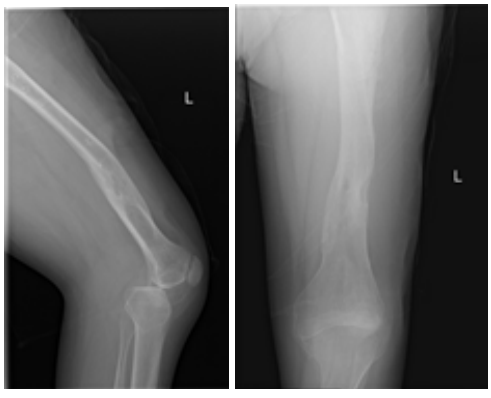


Figure 5: AP x ray of left knee show knee dislocation with posterior translation of tibia

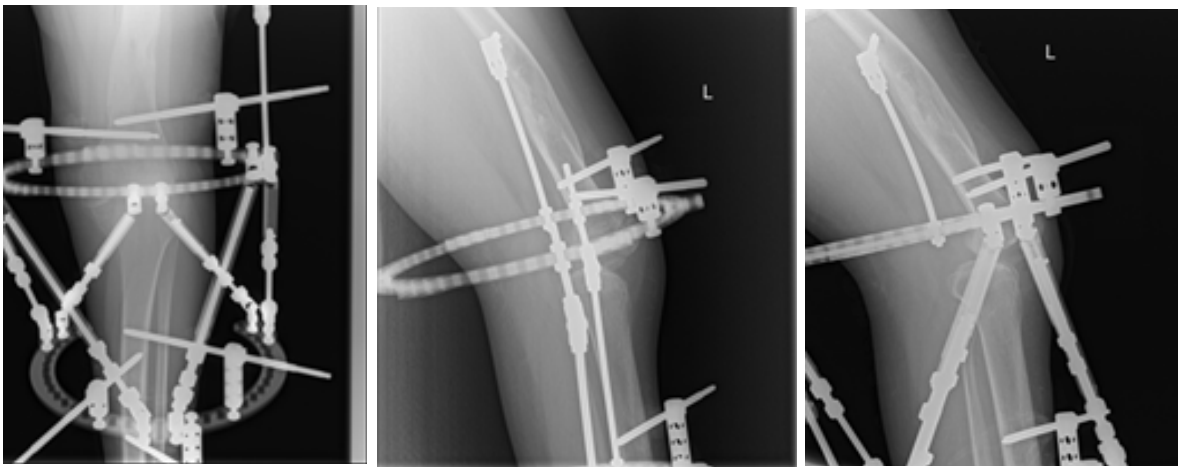


Figure 6: AP x ray of left knee reduce of dislocation with application of TSF

The TSF was kept for almost two months. Then it was removed, and we started gentle ROM exercises followed by progressive ROM exercises. The patient was doing well with good functional status.

At the age of 16 years, the patient had another fall and got a left femur shaft fracture above the site of the osteotomy (Figure 7). He was treated with a left femur anterograde IM nail [Figure 7]. The patient was followed for two years until the complete union was achieved. He was doing good functional activity. The nail was removed due to infection.

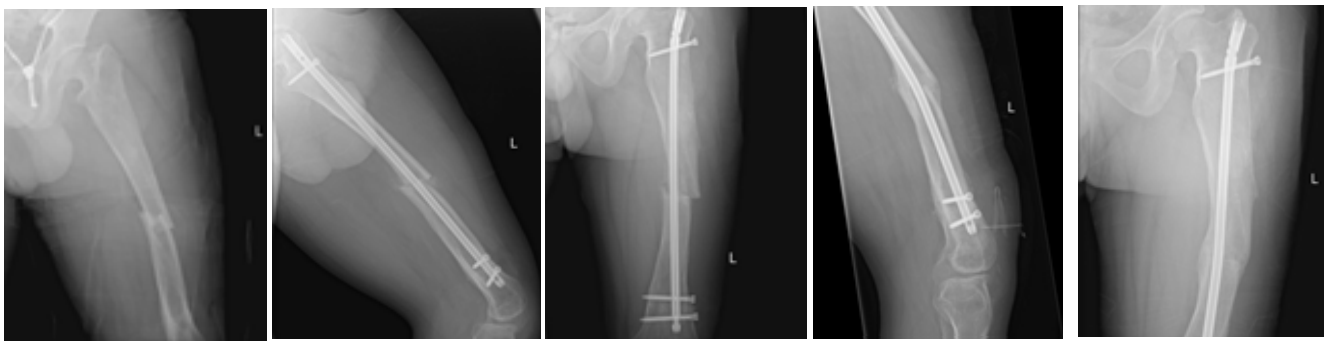


Figure 7: AP and lateral of left femur show fracture of Lt shaft femur transverse fracture. Post left femur anterograde femur nail fixation with mild medial translation and good alignment

After that, the patient complained of chronic knee pain with limited ROM of the affected knee. He could not fully flex and extend his knee. Imaging of the affected knee showed patella hypoplasia (Figure 8). The patient underwent a left knee patellectomy (Figure 9). Follow-up of the patient showed improved ROM. He could achieve full flexion of the left knee post-patellectomy (Figure 10).

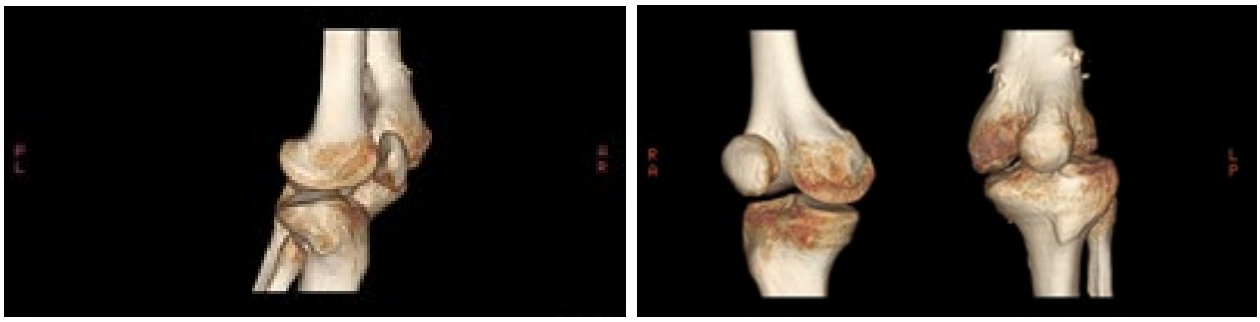


Figure 8: CT sacn 3D of bilateral knee show left patella almost fused with patellofemoral arthritis associated patella hypoplasia. Right patella had normal picture.

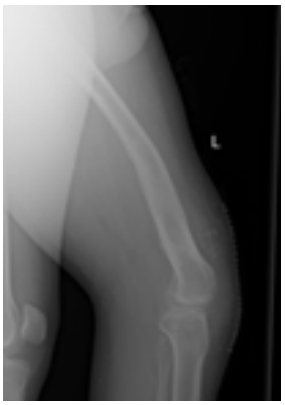


Figure 9: X ray left knee show post patellectomy

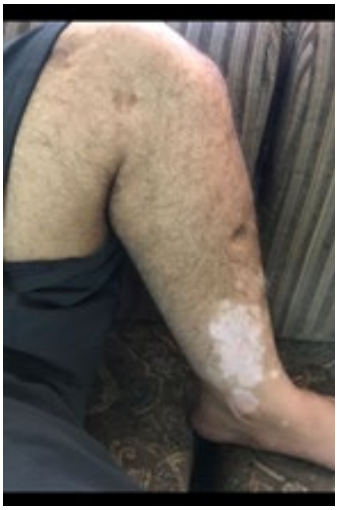


Figure 10: Post left knee patellectomy, full flexion ROM achieved

Moreover, the patient was walking and running in good functional status, following daily activity, full equal length was obtained.

At the age of 24 years, the patient complained of recurrent flexion contracture, with the left knee flexed up to 30 degrees, and he had an extension lag of 15 degrees. We decided to go with left distal femur extension close wedge osteotomy to improve the left knee flexion and extension (Figure 11). The patient was seen post-operation in about three weeks, with full extension and flexion of more than 90 degrees (Figure 12).

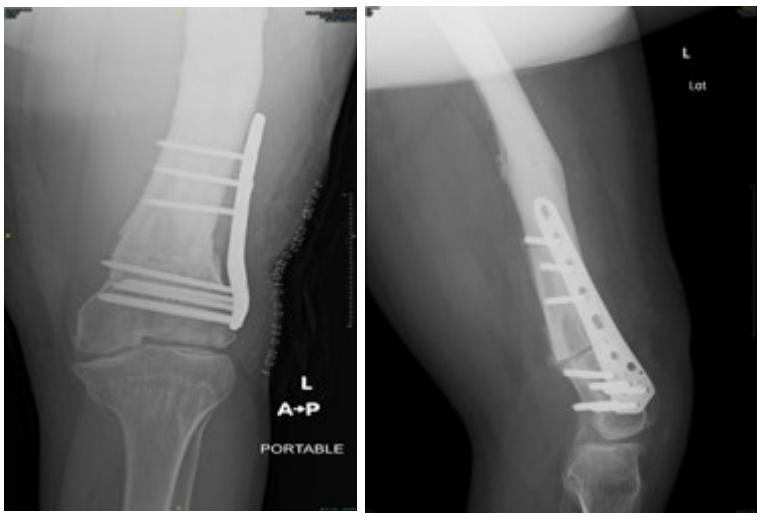


Figure 11: X ray left knee AP lateral view post distal femur extension osteotomy.

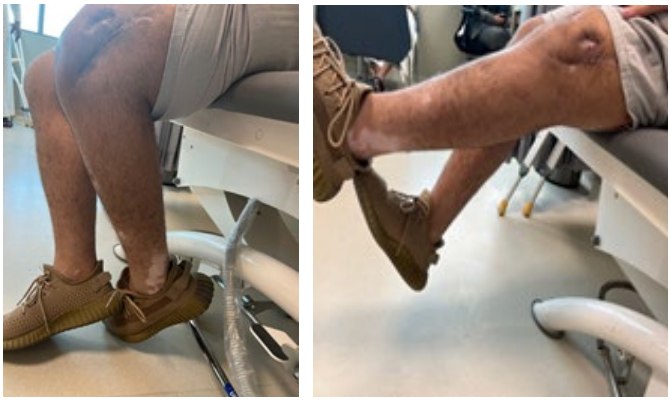


Figure 12: 3 weeks Post left distal femur extension osteotomy, flexion improve from 30 degree to 90-degree, full extension has been achieved

3. Discussion

As was previously mentioned, PFFD is an extremely rare congenital anomaly with few case reports in the literature [1]. Therefore, for us to have one case with a long follow-up that includes five surgeries done to the same patient, which is not described in the existing literature, is exceptional. There are famous risk factors for PFFD. Nearly of the inciting teratogenic mediators for PFFD are hypoxia, ischemia, diabetes mellitus, irradiation, microbiological agents, mechanical injuries, and trauma to the fetus [1]. Amusingly, none of these causes could be linked to our patients or his mothers. From mild cases with minimal femoral shortening and normal hip development to the most severe cases with complete femoral absence, the disorder exhibits a wide clinical spectrum [2].

The affected thigh is usually visibly shorter in the majority of cases, and the hip is frequently flexed, abducted, and externally rotated. Typically, the foot is level with the opposite leg [8.9]. The hip and knee may have flexion contractures, making the limb appear shorter than it actually is anatomically [8]. Our patient exhibited that shortened limb later developed knee flexion contracture with patellofemoral severe arthritis associated with patella hypoplasia. The classification described by Aitken and modified by Amstutz is the one that is most frequently used [1]. classifies PFFD as Class A (femoral head present with varus deformity), Class B (femoral head present with delayed ossification, varus, and pseudoarthrosis may develop), Class C (absent femoral head with acetabular dysplasia and shortening of the femur) and Class D (absent femoral head with severe dysplasia and severe shortening of the femur) [1]. Our patient was classified as type B. Since our patient was older at the presentation (11 years), we made the diagnosis and classification easily with conventional radiography and did not need any further imaging.

Clinical examinations of PFFD cases reveal limb discrepancies with affected thighs that are short and bulky. The severity of femoral deficiency heavily influences the clinical presentations. The lower extremity may even flex, abduct, and rotate externally [2]. The distal femur's deformities at the pelvis include external rotation, neutral to adducted position, and neutral to fixed position. With respect to the proximal femur, the distal femur appears to be extended and adducted to the proximal segment (coxa vara) [8]. The distal femur rotation causes an actual flexion deformity of the distal femur to appear to be an extension, which causes the apparent extension. Flexion contractures of the hip and knee may be present, and the limb may appear shorter than it actually is anatomically [8]. Additionally, a distal femur condyle insufficiency could exist. This is because distal femoral physis is closer to the knee joint on the lateral side. This is often attributed to hypoplasia of the femoral condyle [12]. Our patient exhibited shortening of the affected leg by 6cm to the contralateral leg and limited ROM of the involved hip.

Later, after multiple surgeries, our patient complained of moderate chronic anterior knee pain that was associated with flexion contracture and distal femur condyle deficiency. The course of treatment for a patient with PFFD is determined by the degree of limb length discrepancy, the presence of foot malformations, and the circumstance of the hip and knee joints [2]. PFFD carries a meaningful challenge to efficient management. It needs a multidisciplinary system of prosthetists, pediatric orthopedic surgeons, and physical therapists [1,2]. Therefore, the goals of management will be to balance limb length, correct rotational anomalies, increase pelvic femoral stability, and stabilize the feet [2]. Limb lengthening strategies, such as those used in the present case, should be preferred when the affected limb has been shorted by 50% or when the predicted discrepancy is less than 20 cm [1]. The proximal femur should be fully osseous before lengthening [8]. Treatment frequently requires surgery to allow for the prosthesis to be embedded if the shortness is greater than 50% or the predicted discrepancy is greater than 20 cm [2,8]. Before lengthening, it is important to assess the stability and/or deformity of the hip and knee to evaluate if any surgical procedures needed for the stability of the joint prior lengthening [8].

Since our patient had a stable hip and knee, we decided to go with left femur lengthening with osteotomy and application of TSF. This is because our patient had almost 6 cm shortening compared to the contralateral side. Lengthening osteotomies usually arise in the metaphyseal area of the femur, which is closer to the knee joint and increase force on the knee joint. This surges the risk of knee stiffness and subluxation or dislocation [9,12]. This procedure, which is linked to osteotomy, thus increasing the effective Q angle and increasing the predisposition to lateral subluxation or dislocation of the patella due to bulk of

the quadriceps muscle attached proximally in a lateral position and rotates the knee medially [8,12]. A femur fracture, knee dislocation, and stiffness are the most reported complication post-surgical treatment for PFFD [12]. Our patient faced almost all of them. After lengthening the femur, he had a simple fall down and then had a knee dislocation. After a long time, he again had a simple fall down and ended with a fractured left femur, not on the same site of the osteotomy. The fracture was treated with an IM nail, and after two years, he had the nail removed due to an infected IM nail. After that, our patient complained of flexion contracture and was treated with distal femur extension close wedge osteotomy.

4. Literature review

In a literature review, almost we have reviewed 12 case reports over the last 15 years (since 2008). All reports were taken from PubMed and google scholar. Some cases presented during the intrauterine time with the fetus diagnosed with PFFD. The youngest age of presentation is the age of day one. The most common presentation is related to shortening and lower limb deformity.

Breech presentation, stationary labor, and twin pregnancy are considered risk factors of PFFD. Most radiological findings consist of a deficient proximal left femur, shortening limb, acetabular dysplasia, and absent left femur. Some cases were associated with other dysmorphic features, such as bilateral upper limb meromelia and fibular hemimelia. Only two case reports reported surgical treatment. Moreover, like in our case, only one report staged different surgical treatments of PFFD and its complications.

Reference	Age at presentation (Years)	Symptoms/Signs	Risk factors	Radiology findings	Treatment
Boeisa *et al* This case	11 years	Short left lower limb	None	Shortening left lower limb and acetabular dysplasia	-Lengthening of left femur -Reduction and TSF fixation of a dislocated knee -Left femur IMN for fracture femur -Left patellectomy -Left distal femur extension osteotomy
Felix U. Uduma et al. [1]	4.5 months	Shortened left lower limb Left knee joint flexion contracture	None	Deficient proximal left femur with lateral bowing of its residual shaft. The left femoral capital epiphysis could not be visualized	None
Felix U. Uduma et al. 1	4 months	Short left lower limb noticed from birth. LLD of 4 cm	None	Left lower limb was shortened and in external rotation	None
Özdemir M, et al. 2	20 years	Left lower limb disability Scar tissue in her left thigh due to previous surgeries	None	Left lower limb was about 22cm shorter than its counterpart. Pelvic obliquity Left acetabular dysplasia	Underwent hip release and serial lengthenings between the ages of 2 and 4 years. And pelvic support osteotomy was performed at the age of 12 years. Provided a lower extremity prosthesis during adolescence.
Nana Chunteng et al. 3	1 day	Short thigh Left thigh was bulkier than the right. Limitation of ankle dorsiflexion	Breech presentation and stationery labor.	Shortened left femur with absence of the femoral head.	None
Nana Chunteng et al. 3	2 months	Shortened left lower limb that was noticed at birth.	None	Absent left femur, a dysplastic acetabulum and overriding tibia and fibula	None

Özdemir M. et al. 4	20 years	Bilateral upper extremity and right lower extremity disability Skeletal elements of both upper extremities were absent except the small proximal humeral segments Rt lower limb shortening	None	All skeletal elements of both arms distal to the proximal rudimentary humeral segments were absent . Right lower limb was about 28 cm shorter than its counterpart. Pelvic obliquity and scoliosis	Both upper limb protheses Right hip arthrodesis
Ukkali SB et al. 5	1 day	Short left lower limb Revealed short left lower limb with absence of thigh	None	Ultrasonogram of the left hip joint and lower limb showed absence of proximal femur.	None
Emek Dog˘er et al. 7	20 weeks of gestation	left femur of the fetus was considerably shorter than the right femur, and the length of the left femur was below the fifth percentile	None	Ultrasound images of both the right and left femurs at 20 wg. Left femur: 19.7 mm and the right femur: 31.6 mm were measured.	None
Kubwimana O. 9	3 Y	Short left lower limb noticed from birth.		Left PFFD associated with left fibula hemimelia	None
Shashank Sharma et al. 10	5 Y	shortened limb on the affected side since birth and the deformity had progressed gradually		Hypoplastic proximal femur on the right with lateral bowing of the shaft Fibular hemimelia, deficiency of rays of foot, shortening of tibia and talarequinovalgus	None
Chethan Belgur S et al. 11	4Months	right lower limb shortening		Right-side femoral head and proximal shaft appears deficient resulting in a short femur as compared to normal left-side femur	None

5. Conclusion

Our patient underwent lengthening and correction of PFFD, which has improved his ambulatory status. Definitive treatment of this anomaly is complex, challenging, multidisciplinary, and somewhat difficult to carry out in limited resource facilities. Whichever approach is chosen, the main goal of treatment should always be to improve the patient's ambulatory skills. PFFD needs prolonged treatment. The surgeon must understand how to deal with PFFD and its complications.

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